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Case report

Soft tissue huge solitary neurofibroma in the sacral region without neurofibromatosis: a pediatric case report

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Abstract

Neurofibromas are thick and irregular benign neural sheath tumors touching the peripheral nerve and it may occur at any point along a nerve. Neurofibromas occur frequently as a neurofibromatosis manifestation and less commonly solitary, in unusual sites without neurofibromatosis. The imaging especially MRI is relatively helpful to determine radiological features necessary for the diagnosis, but the definitive diagnosis is established basing on the histopathological examination of the specimen. The primary therapeutic approach for neurofibromas is a complete surgical removal to prevent tumor recurrence. This article highlights a rare case of unusual soft tissue huge solitary neurofibroma in the sacral region without neurofibromatosis in a 12-old-girl.

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Introduction

Neurofibromas are the most common fibroblastic neoplasms affecting penipheral nerves [1]. These benign tumors are originating from different cells constituting the nerve sheath such as neurites, Schwann cells, and fibroblasts. However, the exact cell leading to neurofibroma occurrence has not been obviously identified [2]. Neurofibromas histologic findings vary from collagenous to myxoid matrix according to the neoplastic elements differentiation [1]. Typically, neurofibromas are described in patients with neurofibromatosis. Neurofibromas may be reported as solitary lesions or as part of a generalized syndrome of neurofibromatosis. Rarely, multiple neurofibromas can occur without Von Recklinghausen´s disease. Solitary neurofibroma is a localized tumor that generally occurs in patients without neurofibromatosis as the present case [3,4]. The reported case of soft tissue huge neurofibroma in the sacral region affecting an infant without neurofibromatosis is not well documented in the literature due to the rarity of such tumor at such site.

Patient and observation

A 12-year-old girl was referred to the surgical department for low back painless swelling since the early childhood but neglected by the parents. The patient has unremarkable medical or surgical history. On physical examination, a huge swelling localized in the sacral region was clearly evident on inspection. A palpatory retrosacral mass was found, it was firm, nontender, well limited and measuring about 10*5cm (Figure 1). Neurological examination found no limb paresis. Routine laboratory tests were within normal limits. Magnetic resonance images (Figure 2) showed an 8*7*6cm retrosacral lobulated, non-encapsulated and lobulated mass. The lesion had intermediate signal intensity on T1-weighted images and bright intensity on T2-weighted images, with slight enhancement following the contrast administration. Intraoperatively, a well-circumscribed, encapsulated, firm and white-yellow mass was dissected and a complete excision of the mass was performed with macroscopic adjacent healthy tissue. The resected lesion was ovoid, weighed 300 grams and measured 9*7*6cm (Figure 3). Histopathological report confirmed the diagnosis of neurofibromas and revealed moderate cellularity withelongated and fusiform cells containing several dark-stained nuclei embedded in a collagenous matrix. The post-operative course was uneventful with good esthetic outcomes (Figure 4), and the patient was discharged on the postoperative 3th day with no neurological deficit. A follow-up period of 8 months has shown no evidence of tumor recurrence.

Discussion

Neurofibromas account for 16-30% of all spinal tumors and 13.7% of spinal tumors originating from different elements of nerve sheath including Schwann cells, peri-neurial cells, and fibroblasts. Neurofibromas are most commonly localized in the thoracic region, followed by the cervical and lumbar sites. The sacral region is involved in only 1-5% of all spinal neurofibromas [2]. Neurofibromatosis type I, known as "Von Recklinghausen disease", is a common genetic condition. An autosomal dominant pathology that affects the nervous system related to genetic disorder leading to a defective protein (neurofibromin) which is thought to act as a neoplasm suppressor. Several clinical features characterize the disease such as characterized pigmented skin lesions (café-au-lait macules), benign soft tissues tumors called neurofibromas, distinctive bone lesions and focal iris deformities. Neurofibromas are most commonly associated with neurofibromatosis type I [5]. Neurofibromas are frequently multiple and plexiform with a malignancy potential, they occur as a neurofibromatosis manifestation when they are deep and multiple. Malignant degeneration to neurofibrosarcoma occurs in 4-11% of patients with neurofibromatosis [1,6].

Solitary neurofibromas especially if cutaneous without neurofibromatosis are benign and rarely encountered [1]. The most common clinical presentation for neurofibromas is pain and radiculopathy due to compression of the involved nerve root [5]. Imaging is required for unusual tumor sites, for the tumor extent determination necessary to establish a proper surgical tumor management. On magnetic resonance imaging (MRI), neurofibromas may be inhomogeneous, tend to be isointense with muscle on T1-weighted images and show marked bright intensity with slight enhancement following the contrast administration on T2- weighted images [1,7]. Histologically, all neurofibromas are mixed tumors and are composed of non-neoplastic nerve elements including mast cells fibroblasts and sheath elements [8,9]. The different cellular components of neurofibromas are embedded in an abundant collagenous and often myxoid extracellular matrix containing fibroblasts, Schwann cells, collagen and mucin [10]. The primary therapeutic option for neurofibromas is surgery. These tumors can be surgically removed and the surgical decision should be carefully made basing on the clinical and imaging features of the neoplasm. The complete surgical resection of the lesion is the treatment of choice for neurofibromas. A total lesion removal is necessary to prevent local recurrence and malignant transformation. The neurologic outcome is related to the tumor site and to weather the adjacent nerve root.

Conclusion

This article is dealing with a rare location of neurofibroma with particular features. Surgeons should be aware of this entity especially when the patient does not present neurofibromatosis manifestations. The exact diagnosis would be definitively determined by the histopathological examination of the specimen.

Competing interests

The authors declare no competing interests.

Authors' contributions

All the authors have read and agreed to the final manuscript.

Figures

Figure 1: clinical photo showing the lesion. Retrosacral huge mass measuring about 10x5cm

Figure 2: magnetic resonance images showed a 8x7x6cm retrosacral lobulated, encapsulated and well-limited mass. The lesion shows slight enhancement following the contrast administration

Figure 3: the resected lesion was ovoid, weighed 300 grams and measured 9x7x6cm

Figure 4: post-operative appearance showing esthetic result

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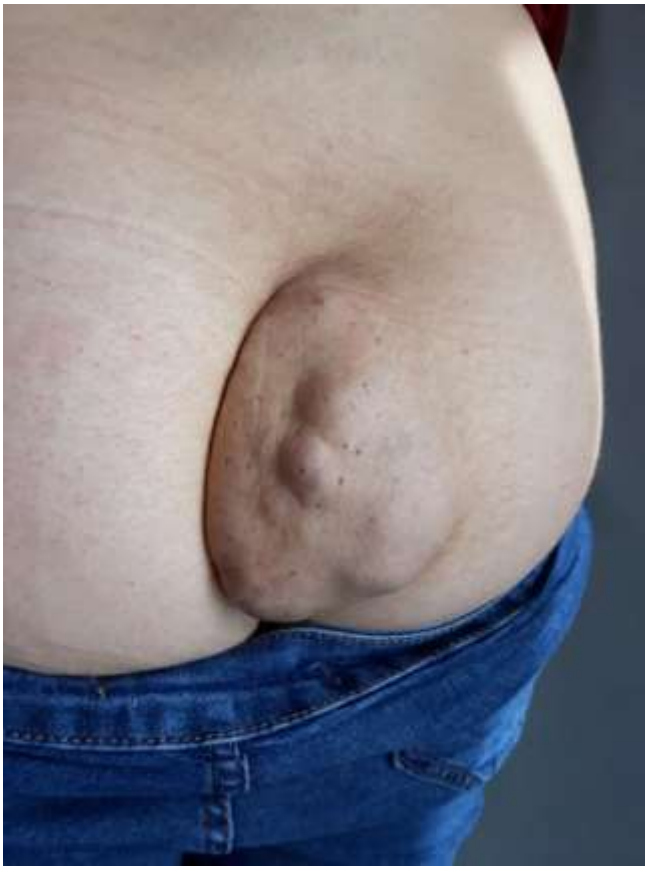

Figure 1: clinical photo showing the lesion. Retrosacral huge mass measuring about 10x5cm

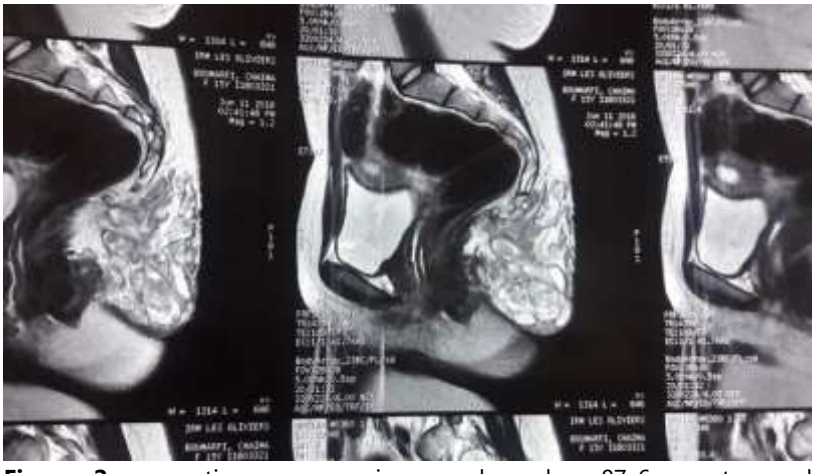

Figure 2: magnetic resonance images showed a 87x6cm retrosacral lobulated, encapsulated and well-limited mass. The lesion shows slight enhancement following the contrast administration

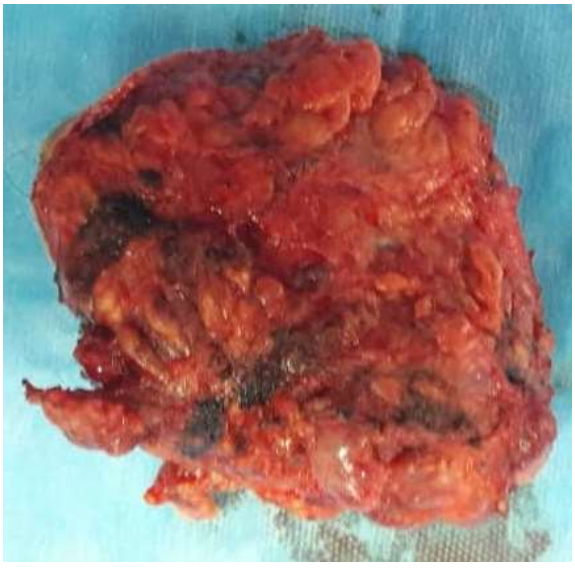

Figure 3: the resected lesion was ovoid, weighed 300 grams and measured 9x7x6cm

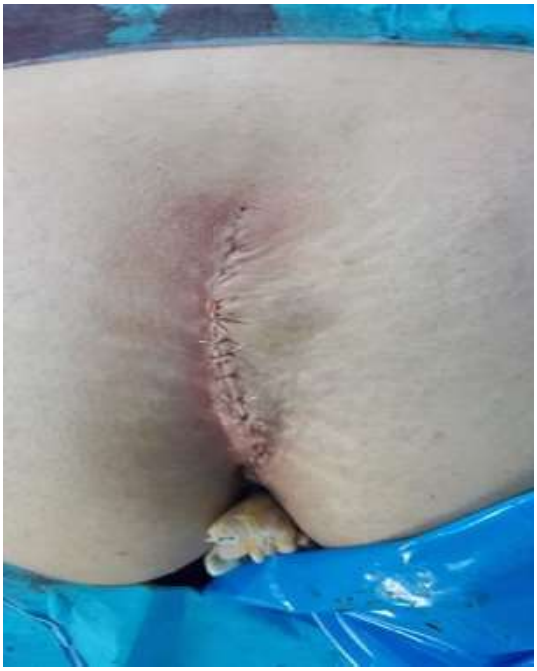

Figure 4: post-operative appearance showing esthetic result